GENE NEWS

A Publication of the Hawai`i Department of Health Genetics Program

Hemoglobinopathy Clinic

Yes, you heard it right here. Genetic services are expanding in the state! A multi-disciplinary Hemoglobinopathy Clinic has become a reality.

Why do we care about hemoglobinopathies?

As you might have guessed, hemoglobinopathies are ***inherited*** blood disorders. This means that they can be passed from parents to child. For this reason, knowing the genetics of the different hemoglobinopathies is important! A correct diagnosis will allow medical professionals to properly manage a patient's condition. Parents will also be able to learn their recurrence risk for future children.

What is a hemoglobinopathy?

A hemoglobinopathy is a blood disorder caused by or associated with the presence of abnormal hemoglobins (oxygen-carrying molecules). Sometimes a person with a hemoglobinopathy will have abnormally shaped red blood cells, while at other times they will not be able to produce enough red blood cells. Red blood cells are important because they carry the oxygen in our bodies. Examples of hemoglobinopathies include sickle cell anemia and thalassemia.

When did the clinic start?

Since June, Hawai`i Community Genetics has been holding a weekly **Hemoglobinopathy Clinic**. Patients are seen by a hematologist (a physician who specializes in blood disorders), a geneticist, and a genetic counselor or genetics nurse.

If you have any questions, please contact

Kirsty McWalter
Hawai`i DOH Genetics Program
(kirsty@hawaiigenetics.org)

To schedule an appointment, please contact

Marissa Short
Hemoglobinopathy
Clinic Coordinator at 973-7303.

Hawai`i Department of Health
Children with Special Health Needs Branch

(808) 733-9055
(808) 733-9068(fax)
www.hawaiigenetics.org

Coordinator's Corner

This issue celebrates the 20th anniversary of the Hawai`i legislation to develop the Newborn Metabolic Screening Program in the Department of Health. It is with great pleasure that we bring you comments and pictures from families who have benefited from newborn screening in Hawai`i. We hope that the family stories show the love and aloha that they want to express to the many people in this state who make newborn screening so successful.

On a less happy note, Allison Taylor has left the Department of Health Genetics Program. However, we are happy that she is still in Hawai`i working at the Queen's Medical Center. Allison is expanding their cancer genetic counseling program. We wish her all the best in her new job.

If you have further questions or comments, please contact me.

Sylvia Au, MS, CGC
sylvia@hawaiigenetics.org
(808) 733-9063

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20 Years of Newborn Screening

Did You Know?

- Over 14 million babies receive newborn screening each year in the U.S.
- Every day, seven babies are born in the U.S. with disorders detectable through newborn screening.
- Every state in the U.S. has a newborn screening program.
- One of the disorders that is included in the Hawai`i newborn screening panel is believed to be responsible for over 200 cases of SIDS (Sudden Infant Death Syndrome) per year.
- Approximately 99.7% of all babies born in Hawai`i receive newborn screening each year

Hawai`i Newborn Screening Program Staff
From left: Loretta Freitas, Christine Matsumoto & Jan Kong

Newborn screening identifies disorders in newborns early to prevent serious medical crises, mental retardation, and death. The screening results also provide important health information to at-risk families.

In 1985, the Hawai`i State Legislature passed legislation to create the Hawai'i Newborn Metabolic Screening Program (NBMSP). Almost a decade later, in 1996, the Legislature passed an amendment to the legislation allowing the NBMSP to expand the number of disorders in the panel and be self-sustaining with user fees and a special fund. Then two years ago, Hawai`i was one of the first states in the nation to expand the statewide screening panel to 31 disorders.

An amazing number (99.7%) of all eligible newborns receive newborn screening in Hawai`i. The success of newborn screening in our state is due to the many individuals and organizations working together over the past twenty years. We applaud the many nurses, doctors, lab workers, hospital administrators, insurers, public health staff and families who give their time and energy to ensure that Hawai`i newborns receive timely and accurate newborn screening.

On the 20th anniversary of the legislation to create the Hawai`i NBMSP, families who have benefited from newborn screening want to share their thanks with everyone who make newborn screening such a successful program.

"I cannot begin to imagine the kind of existence our family would have, had it not been for the newborn screening program. The quality of our entire life and that of our daughter would have been permanently and irreversibly affected."

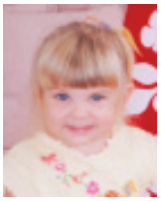
Parents of Maiya, age 6

"Finding out early that our daughter was born with Maple Syrup Urine Disease probably saved her a life of possible retardation, multiple complications and death. We have been to a couple of MSUD symposiums and have seen first hand what kind of life my daughter would have had if she was not picked up within the first few days of her life. We are eternally grateful.

Parents of Jaylee, age 6

"We'd like to think that newborn screening is the reason our daughter is above average in all her development. Our family has benefited from this program greatly. Thank you very much!

Parents of Samantha, age 3



"My life would have been totally different without the newborn screening. I am grateful to the State of Hawai`i for the policies on newborn screening and the additional support I received through Children with Special Health Needs Branch."

Parents of Sarah, age 7

Disorders Detected

PKU	**14**
CH	**107**
CAH	**6**
MSUD	**5**
BIO	**4**
HB	**5**
MCADD	**1**
CPT 1	**3**
HCSD	**1**

PKU phenylketonuria
CH congenital hypothyroidism
CAH congenital adrenal hyperplasia
MSUD maple syrup urine disease
BIO biotinidase deficiency.
HB hemoglobinopathies (sickle cell disease)
MCADD medium chain acyl-CoA dehydrogenase deficiency
CPT 1 carnitine palmitoyl transferase deficiency-type 1
HCSD holocarboxylase synthetase deficiency

Since the beginning of the NBMSP, 146 children with confirmed disorders and over 4,000 at-risk families have been found by newborn screening.

"We feel so fortunate to be in a state that provides this screening. This testing avoids tragedy, struggle and further, on a financial note, saves the taxpayers the cost of health and education demands."

Parents of Matthew, age 7

"Had my daughter not received immediate care, she could have suffered a mental disability. My daughter has maintained great health due to her medication, thanks to the newborn screening test finding her abnormality."

Parents of Dina, age 2

"Thanks to the Newborn Screening test, we were able to start our son on hormone therapy right away and we're proud to say that our son is a healthy five year old."

Parents of Kauiki, age 5

"We remain forever grateful that the State of Hawai`i tests for over 31 disorders. We had moved here from California which only tests for 4 disorders. Thanks to newborn screening, our son has a chance to grow up to be a kind and productive member of society who can make a positive difference in the world. He is our first-born and we love him so much. We would have been devastated if he had died and it could have easily been prevented."

Parents of Gabriel, 5 months

"We are so grateful for newborn screening. If we didn't have newborn screening, we would not have known something was wrong until later in life."

Parents of Ivory, age 6

"I am so thankful that when I delivered my first baby, the metabolic condition that my baby has was included in the newborn screening that other states don't have. Now, my son is five years old and I also have a 3 year old girl with the same condition. They are doing well and are both normal kids."

Mother of Jowell & Lauren, ages 5 & 3

For more information, go to ***www.hawaiigenetics.org.***

Hawai`i Department of Health
Genetics Program
741 Sunset Avenue
Honolulu, HI 96816

Don't Let Newborn Screening Results Fool You

Newborn screening has been credited with saving thousands of children from mental retardation or death, and it is routinely performed in every state in the U.S. **However, due to its great reputation, many people often forget that newborn screening has its limits.**

Why are we telling you this?

We want to remind you that **any baby with symptoms of a disorder in the newborn screening panel needs diagnostic testing to confirm or rule out the diagnosis**, regardless of the screening results.

It's true that a normal newborn screen makes it less likely that a baby has any of the disorders tested for. However, a normal result does not eliminate this possibility, and it should not prevent a baby from receiving a diagnostic test. Although they are rare, false negative test results (when a baby has a normal screening result despite having a disorder) do occur.

Why do false negative results occur?

Newborn screening was designed to identify classic cases of a disorder, so it may miss disorder variants. Premature infants also tend to have a higher number of false negative results than infants born at term. It is therefore recommended that the newborn screen be repeated when the premature infant is a little older. Finally, the slight biological and chemical differences between babies may affect the screening results.

The bottom line:

If you think a baby is showing symptoms of a certain disorder, order the diagnostic tests!